



THE CALIFORNIA

# EXPANDED AFP SCREENING PROGRAM

BASIC BOOKLET FOR WOMEN  
UNDER 35 YEARS OF AGE



There is a different booklet for  
**women 35 years and older.**  
These women should ask  
their doctor or clinic for  
the green booklet called:  
**“Choices in Prenatal Testing  
for Women 35 Years and Older.”**

**The California Expanded AFP Screening Program is voluntary.  
Women can refuse testing without losing eligibility  
or services from State programs.**

CALIFORNIA DEPARTMENT OF HEALTH SERVICES

**Genetic Disease Branch**

2151 Berkeley Way, Annex 4

Berkeley, CA 94704

(510) 540-2534

# *The California Expanded AFP Screening Program*

Every pregnant woman wonders about the health of her fetus (unborn baby) and the possibility of birth defects. The Expanded AFP blood test can help detect some birth defects. This booklet describes the test **for women under 35 years of age at delivery**. It is a woman's own decision whether to have the test or not. A consent/refusal form is at the end of this booklet.

The Program helps detect open neural tube defects, abdominal wall defects, Down syndrome, and trisomy 18. The Expanded AFP Screening Program consists of: the Expanded AFP blood test first, followed by diagnostic tests if needed.

The test results apply only to this pregnancy.

## **W**ho should consider having the Expanded AFP blood test?

### *All pregnant women.*

This blood test is the best way to find out if this pregnancy has a high or low risk of certain birth defects.

Some women may need genetic counseling before deciding about this test. If a woman (or the baby's father) has a medical or family history of inherited conditions, she should discuss the test with her doctor. A woman with a high risk pregnancy should also talk to her doctor. There may be special tests that should be done for these women.

## What does the blood screening test involve?

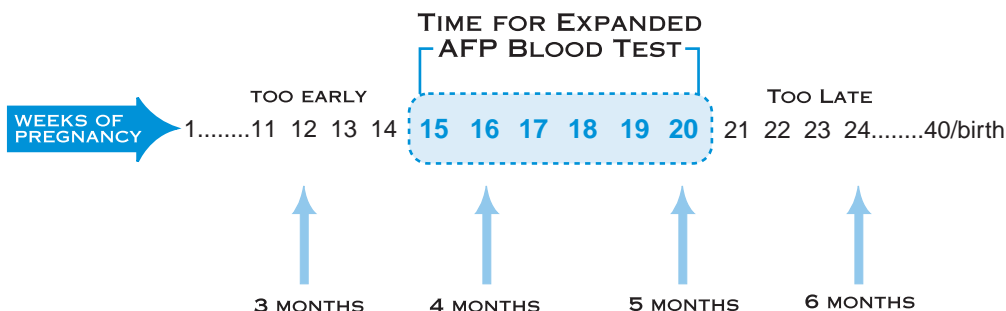
A small amount of blood is taken from the pregnant woman's arm. Her blood is tested for the amount of **AFP** (alpha-fetoprotein), **HCG** (human chorionic gonadotropin), and **UE** (unconjugated estriol). These substances are made by the mother's placenta and the fetus. At each week of pregnancy there are different amounts of these substances in the mother's blood. (What she eats does not affect these substances.)



## When is the blood screening test done?

The blood test can only be done reliably **between 15 and 20 weeks of pregnancy**. *The best time is 16 to 17 weeks.* It is important to know how far along the pregnancy is. Ultrasound is very useful for this purpose.

The result of the blood test is sent to the patient's doctor or clinic within 1-2 weeks.



## **W**hat does a “screen negative” result mean?

It means that the risk for *certain* birth defects is low enough that the Program does not consider follow-up tests necessary. The risk is calculated by measuring the amounts of AFP, HCG and UE in the woman’s blood and also by considering her age. About 90% of women tested will have a “screen negative” result.

Since the blood test is just a screening test, *there is still a chance that the fetus may have a problem* — even when the result is “screen negative.”

## **W**hat does a “screen positive” result mean?

It means that the risk for certain birth defects in this pregnancy is higher than usual (including neural tube defects, abdominal wall defects, Down syndrome, and trisomy 18). The risk is calculated using the amounts of AFP, HCG and UE found in the woman’s blood. Her age is part of the calculation for the risk of Down syndrome and trisomy 18.

Most of the time, however, the reason for this result is *not* a birth defect. The **most common reasons** for a “screen positive” result include:

- ◆ the due date is earlier or later than thought *or*
- ◆ there is more than one fetus (twins, triplets) *or*
- ◆ the substances in the blood varied more than usual, without any known pregnancy problem.

To determine the reason for the “screen positive” result, **genetic counseling and follow-up diagnostic tests are offered** and paid for by the Program. Since receiving this result may cause anxiety, it is important to remember that *most women with “screen positive” results will have normal follow-up tests and healthy babies.*

### SUMMARY OF TEST RESULTS

“screen negative” →	No follow-up tests are offered by the Program.
“screen positive” →	Follow-up tests are provided at no extra cost at a State-approved Prenatal Diagnosis Center.

## **I**f the test is “screen positive,” what happens then?

A woman with a “screen positive” result will be **called by her doctor or clinic**. She will be offered diagnostic services at a **State-approved Prenatal Diagnosis Center**. When authorized, these are the follow-up services covered by the Program:

- **Genetic counseling** - A professional counselor discusses the pregnancy and family medical history. Questions are answered to help the woman make decisions about further testing.
- **Ultrasound** - A picture of the fetus is made using sound waves. This picture shows the age of the fetus and whether there are twins. The detailed ultrasound done at a Prenatal Diagnosis Center can also detect certain birth defects.
- **Amniocentesis** - A small amount of fluid is taken out of the uterus by experienced, State-approved doctors. The fluid and the fetal cells in it are tested for specific birth defects.

Women may refuse any of these services at any time.

**Please remember:** most women with “screen positive” results will have normal follow-up tests and healthy babies.

## **W**hat if the follow-up tests show that the fetus has a birth defect?

Information will be given to the woman by a doctor or genetic counselor at the Prenatal Diagnosis Center. They will discuss the type of birth defect that has been found and any available treatments. They will also discuss options for continuing or ending the pregnancy. The woman can then make a decision.

The Expanded AFP Screening Program does not pay for any other medical services after the follow-up tests. Referrals for special support services are available.

## Birth Defects Found by the Program

**W**hat birth defects may be found through follow-up testing?

**Open neural tube defects, abdominal wall defects, Down syndrome, trisomy 18,** and some other birth defects may be found.

### Neural Tube Defects (NTDs)

As a fetus is forming, the neural tube extends from the top of the head to the end of the spine. This develops into the baby's brain and spinal cord. The neural tube is completely formed by 5 weeks after conception.

If there is an opening in the spine, it is called **spina bifida**. This defect often causes paralysis of the legs. It may also cause loss of bowel and bladder control. Frequently, there is water-on-the-brain (hydrocephaly) which requires surgery.

**Anencephaly** occurs when most of the brain does not develop. This defect causes the death of the fetus or newborn.



### Abdominal Wall Defects

Fetuses with these defects have **abnormal openings** on the abdomen. Intestines and other organs form outside the body. Surgery after birth often corrects the defect.

## Birth Defects Found by the Program

### Down Syndrome

Down syndrome is a common cause of mental retardation. Heart defects are often present, as well. Down syndrome is caused by an extra chromosome #21. Chromosomes are packages of genetic material found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

Down syndrome can occur in the fetus of a woman of any age. However, as a woman gets older, her chances increase for carrying a fetus with Down syndrome.

### Trisomy 18

**Trisomy 18** is caused by an extra chromosome #18. Babies with trisomy 18 have severe mental retardation and physical defects. They usually die before birth or in early infancy.

### HOW MANY BIRTH DEFECTS ARE FOUND?

These birth defects **do not occur very often**. However, *if there is one of these birth defects*, the Expanded AFP Screening Program helps detect it. Among all women who have the Expanded AFP blood test and follow-up tests, the Program finds:

- 97% of the cases of anencephaly
- 80% of the cases of open spina bifida
- 85% of the cases of abdominal wall defects
- 60% of the cases of trisomy 18
- 40% to 66% of the cases of Down syndrome in the pregnancies of women 35 years of age and under. The percentage for Down syndrome varies with the woman's age.



## **C**an the Expanded AFP Screening Program detect every type of birth defect?

**No.** There are birth defects which **cannot** be detected by Expanded AFP Screening. Even when the blood test is “screen negative,” there is still a chance the fetus may have a problem.

## **H**ow much does the Expanded AFP Screening Program cost?

As of 1998, the fee is **\$105**. (Check with the doctor or clinic about the most current fee.) The fee covers the blood test **and** authorized follow-up services at a State-approved Prenatal Diagnosis Center.

The Program mails a bill to women who have the blood test. Women with private insurance should submit the bill to their insurance company. Women who have a prepaid health plan (HMO) may not receive a bill. If they do, they should send the bill to their health plan office.



**As of January 1, 1999, insurance companies and HMOs are required to pay for Expanded AFP testing.**

Women with Medi-Cal usually do not receive a bill. If they do, they should return the bill with their Medi-Cal number. Women without health insurance may make monthly payments and are responsible for the whole amount.

*Any charges for drawing blood are not included in the program fee.*

If you have questions about the test, ask your doctor.  
After you have decided, please sign the consent/refusal  
form on the next page.

CLINICIAN'S COPY

(Remove and file in patient's chart.)

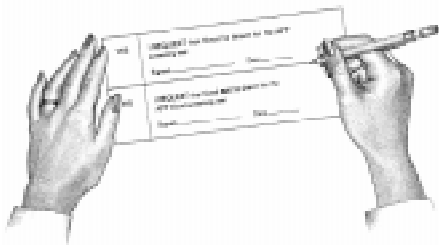
Patient's name \_\_\_\_\_  
(PLEASE PRINT)

ID# \_\_\_\_\_

CONSENT/REFUSAL

FOR THE CALIFORNIA EXPANDED AFP SCREENING PROGRAM

- I have read the information about the **California Expanded AFP Screening Program** which is contained in this booklet (or have had it read to me by \_\_\_\_\_).
- I have been informed that:
  - the purpose of the California Expanded AFP Screening Program is to detect most fetuses with open neural tube defects, abdominal wall defects, Down syndrome, and trisomy 18. However, not all such defects can be detected by the Program.
  - there are other birth defects that cannot be detected by this Program.
  - if the result is "screen positive," I will need to make a decision regarding follow-up testing. Authorized follow-up tests are covered by the Program and will be discussed with me in more detail.
  - if the result is "screen negative," the Program will not pay for any follow-up testing.
  - if the fetus is found to have a birth defect, the decision to continue or terminate the pregnancy will be entirely mine.
  - participation in the California Expanded AFP Screening Program is voluntary. I can refuse any tests at any time.



(over)

- 3. I have read the detection rates for certain birth defects as they are described in this booklet.
- 4. I have been informed that a blood specimen for the California Expanded AFP Screening Program is only reliable between 15 and 20 weeks of pregnancy.
- 5. I have had my questions answered to my satisfaction.

YES	<p>I request that blood be drawn for the Expanded AFP Screening Program.</p> <p>Signed _____ Date _____</p> <p>I should have my blood drawn between</p> <p>_____ and _____</p> <p>month    day    year            month    day    year</p>
No	<p>I request that blood <u>not</u> be drawn for the Expanded AFP Screening Program.</p> <p>Signed _____ Date _____</p>

*I understand that the blood specimen and information obtained during the testing process become the property of the California Department of Health Services. They may be used for program evaluation or research by the Department or Department-approved scientific researchers without identifying the person or persons from whom these results were obtained, unless I specifically prohibit such use in writing. All information procured by the Department of Health Services, or by any other person, agency or organization acting jointly with the Department in connection with such special studies, shall be confidential. I may obtain additional information about the study or prohibit the use of my specimen by writing George Cunningham, MD, MPH, Genetic Disease Branch, 2151 Berkeley Way, Annex 4, Berkeley, CA 94704.*

*If new information becomes available about a birth defect detected during this pregnancy, the information may be sent to me unless I specifically prohibit it by writing to George Cunningham, MD, MPH at the above address.*

## PATIENT'S COPY

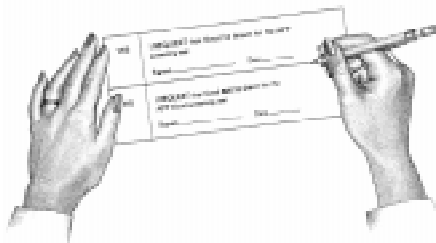
Patient's name \_\_\_\_\_  
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ID# \_\_\_\_\_

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**Please keep this booklet and your copy of the  
consent/refusal form for your records.**



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